

Complete Summary

GUIDELINE TITLE

Prenatal screening and diagnosis for pediatricians.

BIBLIOGRAPHIC SOURCE(S)

Cunniff C. Prenatal screening and diagnosis for pediatricians. Pediatrics 2004 Sep; 114(3):889-94. [42 references] [PubMed](#)

GUIDELINE STATUS

This is the current release of the guideline.

American Academy of Pediatrics (AAP) Policies are reviewed every 3 years by the authoring body, at which time a recommendation is made that the policy be retired, revised, or reaffirmed without change. Until the Board of Directors approves a revision or reaffirmation, or retires a statement, the current policy remains in effect.

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SCOPE

DISEASE/CONDITION(S)

Prenatal genetic disorders (diagnosed or suspected) that may result in fetal abnormalities

GUIDELINE CATEGORY

Counseling

CLINICAL SPECIALTY

Family Practice
Medical Genetics
Obstetrics and Gynecology
Pediatrics

INTENDED USERS

Physicians

GUIDELINE OBJECTIVE(S)

To update the pediatrician about indications for prenatal diagnosis, current techniques used for prenatal diagnosis, and the status of maternal screenings for detection of fetal abnormalities

TARGET POPULATION

- Family members of a child with a birth defect or genetic disorder who may be a risk for recurrence of the same or similar problems in future offspring
- Families in which prenatal diagnosis is being considered or in which there is a fetus with a genetic disorder

INTERVENTIONS AND PRACTICES CONSIDERED

1. Patient education and counseling by a pediatrician regarding current techniques used for prenatal diagnoses
2. Referrals to appropriate specialists

MAJOR OUTCOMES CONSIDERED

- Risk factors for fetal abnormalities
- Risks and benefits of diagnostic techniques

METHODOLOGY

METHODS USED TO COLLECT/SELECT EVIDENCE

Searches of Electronic Databases

DESCRIPTION OF METHODS USED TO COLLECT/SELECT THE EVIDENCE

Not stated

NUMBER OF SOURCE DOCUMENTS

Not stated

METHODS USED TO ASSESS THE QUALITY AND STRENGTH OF THE EVIDENCE

Not stated

RATING SCHEME FOR THE STRENGTH OF THE EVIDENCE

Not applicable

METHODS USED TO ANALYZE THE EVIDENCE

Review

DESCRIPTION OF THE METHODS USED TO ANALYZE THE EVIDENCE

Not stated

METHODS USED TO FORMULATE THE RECOMMENDATIONS

Not stated

RATING SCHEME FOR THE STRENGTH OF THE RECOMMENDATIONS

Not applicable

COST ANALYSIS

A formal cost analysis was not performed and published cost analyses were not reviewed.

METHOD OF GUIDELINE VALIDATION

Peer Review

DESCRIPTION OF METHOD OF GUIDELINE VALIDATION

Not stated

RECOMMENDATIONS

MAJOR RECOMMENDATIONS

Pediatricians may be called on to counsel a family in which prenatal diagnosis is being considered or in which there is a fetus with a genetic disorder. It is important that pediatricians involve themselves at a level appropriate to their training and experience, that they clarify their role in the prenatal diagnostic process with the family, and that they document their discussion and recommendations. In most circumstances, pediatricians will not assume a primary role in performing prenatal diagnostic procedures or counseling the family about their risks and benefits. More frequently, an obstetrician, maternal-fetal medicine specialist, clinical geneticist, and/or a genetic counselor will direct the diagnostic evaluation and provide pretest and posttest counseling. Because of a previous

relationship with the family, the pediatrician may be called on to review this information and assist the family in the decision-making process. The pediatrician should be familiar with the principles of prenatal genetic diagnosis and know how to apply them to specific problems in genetic counseling, diagnosis, and management in clinical practice. Pediatricians should be familiar with resources available in their region for obtaining information about whether and how a specific disorder can be diagnosed and when and where to refer patients for prenatal genetic diagnosis. The technology of prenatal diagnosis is changing rapidly, and genetic consultants can assist pediatricians in the appropriate use and interpretation of the diagnostic tests that are available.

CLINICAL ALGORITHM(S)

None provided

EVIDENCE SUPPORTING THE RECOMMENDATIONS

TYPE OF EVIDENCE SUPPORTING THE RECOMMENDATIONS

The type of evidence supporting each recommendation is not specifically stated.

BENEFITS/HARMS OF IMPLEMENTING THE GUIDELINE RECOMMENDATIONS

POTENTIAL BENEFITS

This guideline may improve pediatrician's familiarity with indications for prenatal diagnosis, current techniques used for prenatal diagnosis, and the status of maternal screenings for detection of fetal abnormalities.

POTENTIAL HARMS

Not stated

QUALIFYING STATEMENTS

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The guidance in this report does not indicate an exclusive course of treatment or serve as a standard of medical care. Variations, taking into account individual circumstances, may be appropriate.

IMPLEMENTATION OF THE GUIDELINE

DESCRIPTION OF IMPLEMENTATION STRATEGY

An implementation strategy was not provided.

INSTITUTE OF MEDICINE (IOM) NATIONAL HEALTHCARE QUALITY REPORT CATEGORIES

IOM CARE NEED

Staying Healthy

IOM DOMAIN

Effectiveness

Patient-centeredness

IDENTIFYING INFORMATION AND AVAILABILITY

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ADAPTATION

Not applicable: The guideline was not adapted from another source.

DATE RELEASED

2004 Sep

GUIDELINE DEVELOPER(S)

American Academy of Pediatrics - Medical Specialty Society

SOURCE(S) OF FUNDING

American Academy of Pediatrics

GUIDELINE COMMITTEE

Committee on Genetics

COMPOSITION OF GROUP THAT AUTHORED THE GUIDELINE

Committee on Genetics, 2003–2004: G. Bradley Schaefer, MD (Chairperson); Marilyn J. Bull, MD; Joseph H. Hersh, MD; Celia I. Kaye, MD, PhD; Nancy J. Mendelsohn, MD; John B. Moeschler, MD; Howard M. Saal, MD

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FINANCIAL DISCLOSURES/CONFLICTS OF INTEREST

Not stated

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GUIDELINE AVAILABILITY

Electronic copies: Available from the [American Academy of Pediatrics \(AAP\) Policy Web site](#).

Print copies: Available from American Academy of Pediatrics, 141 Northwest Point Blvd., P.O. Box 927, Elk Grove Village, IL 60009-0927.

AVAILABILITY OF COMPANION DOCUMENTS

None available

PATIENT RESOURCES

None available

NGC STATUS

This NGC summary was completed by ECRI on October 18, 2004. The information was verified by the guideline developer on November 23, 2004.

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